



## Charcot-Marie-Tooth disease

Charcot-Marie-Tooth disease is a group of progressive disorders that affect the peripheral nerves. Peripheral nerves connect the brain and spinal cord to muscles and to sensory cells that detect sensations such as touch, pain, heat, and sound. Damage to the peripheral nerves can result in loss of sensation and wasting (atrophy) of muscles in the feet, legs, and hands.

Charcot-Marie-Tooth disease usually becomes apparent in adolescence or early adulthood, but onset may occur anytime from early childhood through late adulthood. Symptoms of Charcot-Marie-Tooth disease vary in severity, even among members of the same family. Some people never realize they have the disorder, but most have a moderate amount of physical disability. A small percentage of people experience severe weakness or other problems which, in rare cases, can be life-threatening. In most affected individuals, however, Charcot-Marie-Tooth disease does not affect life expectancy.

Typically, the earliest symptoms of Charcot-Marie-Tooth disease involve balance difficulties, clumsiness, and muscle weakness in the feet. Affected individuals may have foot abnormalities such as high arches (pes cavus), flat feet (pes planus), or curled toes (hammer toes). They often have difficulty flexing the foot or walking on the heel of the foot. These difficulties may cause a higher than normal step (or gait) and increase the risk of ankle injuries and tripping.

As the disease progresses, muscles in the lower legs usually weaken, but leg and foot problems rarely require the use of a wheelchair. Affected individuals may also develop weakness in the hands, causing difficulty with daily activities such as writing, fastening buttons, and turning doorknobs. People with this disorder typically experience a decreased sensitivity to touch, heat, and cold in the feet and lower legs, but occasionally feel aching or burning sensations. In some cases, affected individuals experience gradual hearing loss, deafness, or loss of vision.

There are several types of Charcot-Marie-Tooth disease. Type 1 Charcot-Marie-Tooth disease (CMT1) is characterized by abnormalities in myelin, the fatty substance that covers nerve cells, protecting them and helping to conduct nerve impulses. These abnormalities slow the transmission of nerve impulses. Type 2 Charcot-Marie-Tooth disease (CMT2) is characterized by abnormalities in the fiber, or axon, that extends from a nerve cell body and transmits nerve impulses. These abnormalities reduce the strength of the nerve impulse. Type 4 Charcot-Marie-Tooth disease (CMT4) affects either the axon or myelin and is distinguished from the other types by its pattern of inheritance. In intermediate forms of Charcot-Marie-Tooth disease, the nerve impulses are both slowed and reduced in strength, probably due to abnormalities in both axons

and myelin. Type X Charcot-Marie-Tooth disease (CMTX) is caused by mutations in a gene on the X chromosome, one of the two sex chromosomes. Within the various types of Charcot-Marie-Tooth disease, subtypes (such as CMT1A, CMT1B, CMT2A, CMT4A, and CMTX1) are distinguished by the specific gene that is altered.

Sometimes other, more historical names are used to describe this disorder. For example, Roussy-Levy syndrome is a form of Charcot-Marie-Tooth disease defined by the additional feature of rhythmic shaking (tremors). Dejerine-Sottas syndrome is a term sometimes used to describe a severe, early childhood form of Charcot-Marie-Tooth disease; it is also sometimes called Charcot-Marie-Tooth disease type 3 (CMT3). Depending on the specific gene that is altered, this severe, early onset form of the disorder may also be classified as CMT1 or CMT4. CMTX5 is also known as Rosenberg-Chutorian syndrome. Some researchers believe that this condition is not actually a form of Charcot-Marie-Tooth disease. Instead, they classify it as a separate disorder characterized by peripheral nerve problems, deafness, and vision loss.

## **Frequency**

Charcot-Marie-Tooth disease is the most common inherited disorder that involves the peripheral nerves, affecting an estimated 150,000 people in the United States. It occurs in populations worldwide with a prevalence of about 1 in 2,500 individuals.

## **Genetic Changes**

Charcot-Marie-Tooth disease is caused by mutations in many different genes. These genes provide instructions for making proteins that are involved in the function of peripheral nerves in the feet, legs, and hands. The gene mutations that cause Charcot-Marie-Tooth disease affect the function of the proteins in ways that are not fully understood; however, they likely impair axons, which transmit nerve impulses, or affect the specialized cells that produce myelin. As a result, peripheral nerve cells slowly lose the ability to stimulate the muscles and to transmit sensory signals to the brain.

The list of genes associated with Charcot-Marie-Tooth disease continues to grow as researchers study this disorder. Different mutations within a particular gene may cause signs and symptoms of differing severities or lead to different types of Charcot-Marie-Tooth disease.

CMT1 is caused by mutations in the following genes: *PMP22* (CMT1A and CMT1E), *MPZ* (CMT1B), *LITAF* (CMT1C), *EGR2* (CMT1D), and *NEFL* (CMT1F).

CMT2 can result from alterations in many genes, including *MFN2* and *KIF1B* (CMT2A); *RAB7A* (CMT2B); *LMNA* (CMT2B1); *TRPV4* (CMT2C); *BSCL2* and *GARS* (CMT2D); *NEFL* (CMT2E); *HSPB1* (CMT2F); *MPZ* (CMT2I and CMT2J); *GDAP1* (CMT2K); and *HSPB8* (CMT2L). Certain *DNM2* gene mutations also cause a form of CMT2.

CMT4 is caused by mutations in the following genes: *GDAP1* (CMT4A), *MTMR2* (CMT4B1), *SBF2* (CMT4B2), *SH3TC2* (CMT4C), *NDRG1* (CMT4D), *EGR2* (CMT4E), *PRX* (CMT4F), *FGD4* (CMT4H), and *FIG4* (CMT4J).

Intermediate forms of the disorder can be caused by alterations in genes including *DNM2*, *MPZ*, *YARS*, and *GDAP1*. CMTX is caused by mutations in genes including *GJB1* (CMTX1) and *PRPS1* (CMTX5). Mutations in additional genes, some of which have not been identified, also cause various forms of Charcot-Marie-Tooth disease.

## Inheritance Pattern

The pattern of inheritance varies with the type of Charcot-Marie-Tooth disease. CMT1, most cases of CMT2, and most intermediate forms are inherited in an autosomal dominant pattern. This pattern of inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one affected parent.

CMT4, a few CMT2 subtypes, and some intermediate forms are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but do not show signs and symptoms of the condition.

CMTX is inherited in an X-linked dominant pattern. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome. The inheritance is dominant if one copy of the altered gene is sufficient to cause the condition. In most cases, affected males, who have the alteration on their only copy of the X chromosome, experience more severe symptoms of the disorder than females, who have two X chromosomes. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons. All daughters of affected men will have one altered X chromosome, but they may only have mild symptoms of the disorder.

Some cases of Charcot-Marie-Tooth disease result from a new mutation and occur in people with no history of the disorder in their family.

## Other Names for This Condition

- Charcot-Marie-Tooth hereditary neuropathy
- Charcot-Marie-Tooth syndrome
- CMT
- hereditary motor and sensory neuropathy
- HMSN
- peroneal muscular atrophy
- PMA

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Charcot-Marie-Tooth disease  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0007959/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease and deafness  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1861669/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease dominant intermediate 3  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843075/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2B1  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1854154/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2B2  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1854150/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2C  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2079540/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2D  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1832274/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2E  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843225/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2F  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1847823/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2I  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843251/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2J  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843153/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2K  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842983/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease type 2P  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3280797/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, axonal, type 2b  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1833219/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, axonal, type 2O  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3280220/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, autosomal recessive  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843183/>

- Genetic Testing Registry: Charcot-Marie-Tooth disease, demyelinating, type 1b  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0270912/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, demyelinating, type 1d  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843247/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, demyelinating, type 1f  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843164/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, dominant intermediate C  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842237/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, dominant intermediate E  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3280845/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, recessive intermediate A  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842197/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 1C  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0270913/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 2A1  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1861678/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 2A2  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1836485/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 2L  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1837552/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 2N  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750090/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 4A  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1859198/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 4B1  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1832399/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 4B2  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858278/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 4C  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1866636/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 4D  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1832334/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 4H  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1836336/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type 4J  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1970011/>

- Genetic Testing Registry: Charcot-Marie-Tooth disease, type I  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0751036/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, type IA  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0270911/>
- Genetic Testing Registry: Charcot-Marie-Tooth disease, X-linked recessive, type 5  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1839566/>
- Genetic Testing Registry: Congenital hypomyelinating neuropathy  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0393818/>
- Genetic Testing Registry: Dejerine-Sottas disease  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0011195/>
- Genetic Testing Registry: DNM2-related intermediate Charcot-Marie-Tooth neuropathy  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1847902/>
- Genetic Testing Registry: Roussy-Lévy syndrome  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0205713/>
- Genetic Testing Registry: X-linked hereditary motor and sensory neuropathy  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0393808/>

#### Other Diagnosis and Management Resources

- GeneReview: Charcot-Marie-Tooth Hereditary Neuropathy Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1358>
- GeneReview: Charcot-Marie-Tooth Neuropathy Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1205>
- GeneReview: Charcot-Marie-Tooth Neuropathy Type 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1285>
- GeneReview: Charcot-Marie-Tooth Neuropathy Type 2A  
<https://www.ncbi.nlm.nih.gov/books/NBK1511>
- GeneReview: Charcot-Marie-Tooth Neuropathy Type 2E/1F  
<https://www.ncbi.nlm.nih.gov/books/NBK1187>
- GeneReview: Charcot-Marie-Tooth Neuropathy Type 4  
<https://www.ncbi.nlm.nih.gov/books/NBK1468>
- GeneReview: Charcot-Marie-Tooth Neuropathy Type 4A  
<https://www.ncbi.nlm.nih.gov/books/NBK1539>
- GeneReview: Charcot-Marie-Tooth Neuropathy Type 4C  
<https://www.ncbi.nlm.nih.gov/books/NBK1340>

- GeneReview: Charcot-Marie-Tooth Neuropathy X Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1374>
- GeneReview: Charcot-Marie-Tooth Neuropathy X Type 5  
<https://www.ncbi.nlm.nih.gov/books/NBK1876>
- GeneReview: DNM2-Related Intermediate Charcot-Marie-Tooth Neuropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK45014>
- GeneReview: GARS-Associated Axonal Neuropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK1242>
- GeneReview: TRPV4-Associated Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK201366>
- MedlinePlus Encyclopedia: Charcot-Marie-Tooth Disease  
<https://medlineplus.gov/ency/article/000727.htm>
- MedlinePlus Encyclopedia: Hammer Toe  
<https://medlineplus.gov/ency/article/001235.htm>
- MedlinePlus Encyclopedia: High Arch  
<https://medlineplus.gov/ency/article/001261.htm>

#### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

#### **Additional Information & Resources**

##### MedlinePlus

- Encyclopedia: Charcot-Marie-Tooth Disease  
<https://medlineplus.gov/ency/article/000727.htm>
- Encyclopedia: Hammer Toe  
<https://medlineplus.gov/ency/article/001235.htm>
- Encyclopedia: High Arch  
<https://medlineplus.gov/ency/article/001261.htm>

- Health Topic: Charcot-Marie-Tooth Disease  
<https://medlineplus.gov/charcotmarietoothdisease.html>
- Health Topic: Peripheral Nerve Disorders  
<https://medlineplus.gov/peripheralnervedisorders.html>

#### Genetic and Rare Diseases Information Center

- Charcot-Marie-Tooth disease  
<https://rarediseases.info.nih.gov/diseases/6034/charcot-marie-tooth-disease>
- Charcot-Marie-Tooth disease type 1A  
<https://rarediseases.info.nih.gov/diseases/1245/charcot-marie-tooth-disease-type-1a>
- Charcot-Marie-Tooth disease type 2B  
<https://rarediseases.info.nih.gov/diseases/9192/charcot-marie-tooth-disease-type-2b>
- Charcot-Marie-Tooth disease type 2F  
<https://rarediseases.info.nih.gov/diseases/9194/charcot-marie-tooth-disease-type-2f>
- Roussy Levy syndrome  
<https://rarediseases.info.nih.gov/diseases/4741/roussy-levy-syndrome>

#### Additional NIH Resources

- National Human Genome Research Institute  
<https://www.genome.gov/11009201/>
- National Institute of Neurological Disorders and Stroke  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Charcot-Marie-Tooth-Disease-Information-Page>

#### Educational Resources

- Disease InfoSearch: Charcot-Marie-Tooth Disease  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease/1276>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 1A  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+1A/1284>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 1B  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+1B/1285>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 1C  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+1C/1286>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 1D  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+1D/1287>

- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 1E  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+1E/1288>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 1F  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+1F/1289>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2A  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2A/1290>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2B  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2B/1291>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2C  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2C/1294>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2D  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2D/1295>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2E  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2E/1296>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2F  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2F/1297>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2H  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2H/1299>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 2I  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+2I/1300>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 4A  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+4A/1303>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 4B1  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+4B1/1304>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 4C  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+4C/1307>
- Disease InfoSearch: Charcot-Marie-Tooth Disease Type 4E  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+Type+4E/1308>
- Disease InfoSearch: Charcot-Marie-Tooth Disease X-Linked 1  
<http://www.diseaseinfosearch.org/Charcot-Marie-Tooth+Disease+X-Linked+1/1311>
- Disease InfoSearch: Hereditary motor and sensory neuropathy with optic atrophy  
<http://www.diseaseinfosearch.org/Hereditary+motor+and+sensory+neuropathy+with+optic+atrophy/8523>
- Johns Hopkins Medicine  
[http://www.hopkinsmedicine.org/neurology\\_neurosurgery/centers\\_clinics/peripheral\\_nerve/conditions/charcot\\_marie\\_tooth\\_disease.html](http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/peripheral_nerve/conditions/charcot_marie_tooth_disease.html)

- MalaCards: charcot-marie-tooth disease  
[http://www.malacards.org/card/charcot\\_marie\\_tooth\\_disease](http://www.malacards.org/card/charcot_marie_tooth_disease)
- Merck Manual Consumer Version  
<http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/peripheral-nerve-disorders/charcot-marie-tooth-disease>
- My46 Trait Profile  
<https://www.my46.org/trait-document?trait=Charcot%20Marie%20Tooth%20disease&type=profile>
- National Health Service (UK)  
<http://www.nhs.uk/Conditions/Charcot-Marie-Tooth-disease/Pages/Introduction.aspx>
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=64746](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=64746)
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2C  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99937](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99937)
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2D  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99938](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99938)
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2E  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99939](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99939)
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2F  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99940](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99940)
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2I  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99942](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99942)
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2L  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99945](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99945)
- Orphanet: Autosomal dominant Charcot-Marie-Tooth disease type 2N  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=228174](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=228174)
- Orphanet: Autosomal recessive Charcot-Marie-Tooth disease with hoarseness  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101097](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101097)
- Orphanet: Charcot-Marie-Tooth disease  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=166](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=166)
- Orphanet: Charcot-Marie-Tooth disease type 1  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=65753](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=65753)
- Orphanet: Charcot-Marie-Tooth disease type 1A  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101081](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101081)

- Orphanet: Charcot-Marie-Tooth disease type 1B  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101082](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101082)
- Orphanet: Charcot-Marie-Tooth disease type 1C  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101083](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101083)
- Orphanet: Charcot-Marie-Tooth disease type 1D  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101084](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101084)
- Orphanet: Charcot-Marie-Tooth disease type 1E  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=90658](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=90658)
- Orphanet: Charcot-Marie-Tooth disease type 1F  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101085](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101085)
- Orphanet: Charcot-Marie-Tooth disease type 2B1  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=98856](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98856)
- Orphanet: Charcot-Marie-Tooth disease type 2B2  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101101](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101101)
- Orphanet: Charcot-Marie-Tooth disease type 2H  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=101102](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=101102)
- Orphanet: Charcot-Marie-Tooth disease type 4  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=64749](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=64749)
- Orphanet: Charcot-Marie-Tooth disease type 4A  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99948](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99948)
- Orphanet: Charcot-Marie-Tooth disease type 4B1  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99955](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99955)
- Orphanet: Charcot-Marie-Tooth disease type 4B2  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99956](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99956)
- Orphanet: Charcot-Marie-Tooth disease type 4C  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99949](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99949)
- Orphanet: Charcot-Marie-Tooth disease type 4D  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99950](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99950)
- Orphanet: Charcot-Marie-Tooth disease type 4E  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99951](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99951)
- Orphanet: Charcot-Marie-Tooth disease type 4F  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99952](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99952)
- Orphanet: Charcot-Marie-Tooth disease type 4H  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=99954](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99954)
- Orphanet: Charcot-Marie-Tooth disease type 4J  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=139515](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=139515)

- Orphanet: Dejerine-Sottas syndrome  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=64748](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=64748)
- Orphanet: Dejerine-Sottas syndrome  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=64748](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=64748)
- Orphanet: X-linked Charcot-Marie-Tooth disease  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=64747](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=64747)
- University of Chicago  
<http://peripheralneuropathycenter.uchicago.edu/learnaboutpn/typesofpn/hereditary/charcotmarietooth.shtml>

#### Patient Support and Advocacy Resources

- Charcot-Marie-Tooth Association  
<http://www.cmtausa.org/>
- Hereditary Neuropathy Foundation  
<http://www.hnf-cure.org/>
- Muscular Dystrophy Association  
<https://www.mda.org/disease/charcot-marie-tooth>
- National Organization for Rare Disorders (NORD): Charcot-Marie-Tooth Disease  
<https://rarediseases.org/rare-diseases/charcot-marie-tooth-disease/>
- National Organization for Rare Disorders (NORD): Dejerine Sottas Disease  
<https://rarediseases.org/rare-diseases/dejerine-sottas-disease/>
- National Organization for Rare Disorders: Rosenberg-Chutorian Syndrome  
<https://rarediseases.org/rare-diseases/rosenberg-chutorian-syndrome/>
- National Organization for Rare Disorders: Roussy-Levy Syndrome  
<https://rarediseases.org/rare-diseases/roussy-levy-syndrome/>

#### GeneReviews

- Charcot-Marie-Tooth Hereditary Neuropathy Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1358>
- Charcot-Marie-Tooth Neuropathy Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1205>
- Charcot-Marie-Tooth Neuropathy Type 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1285>
- Charcot-Marie-Tooth Neuropathy Type 2A  
<https://www.ncbi.nlm.nih.gov/books/NBK1511>
- Charcot-Marie-Tooth Neuropathy Type 2E/1F  
<https://www.ncbi.nlm.nih.gov/books/NBK1187>

- Charcot-Marie-Tooth Neuropathy Type 4  
<https://www.ncbi.nlm.nih.gov/books/NBK1468>
- Charcot-Marie-Tooth Neuropathy Type 4A  
<https://www.ncbi.nlm.nih.gov/books/NBK1539>
- Charcot-Marie-Tooth Neuropathy Type 4C  
<https://www.ncbi.nlm.nih.gov/books/NBK1340>
- Charcot-Marie-Tooth Neuropathy X Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1374>
- Charcot-Marie-Tooth Neuropathy X Type 5  
<https://www.ncbi.nlm.nih.gov/books/NBK1876>
- DNM2-Related Intermediate Charcot-Marie-Tooth Neuropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK45014>
- GARS-Associated Axonal Neuropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK1242>
- TRPV4-Associated Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK201366>

#### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?term=%22Charcot-Marie-Tooth+disease%22+%5BDISEASE%5D+OR+NCT00004568+%5BID-NUMBER%5D>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Charcot-Marie-Tooth+Disease%5BMAJR%5D%29+AND+%28Charcot-Marie-Tooth+disease%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

#### OMIM

- CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS  
<http://omim.org/entry/118300>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL DOMINANT, TYPE 2A2A  
<http://omim.org/entry/609260>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2A1  
<http://omim.org/entry/118210>

- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B  
<http://omim.org/entry/600882>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B1  
<http://omim.org/entry/605588>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B2  
<http://omim.org/entry/605589>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2D  
<http://omim.org/entry/601472>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2E  
<http://omim.org/entry/607684>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2F  
<http://omim.org/entry/606595>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2I  
<http://omim.org/entry/607677>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J  
<http://omim.org/entry/607736>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2K  
<http://omim.org/entry/607831>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2L  
<http://omim.org/entry/608673>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2N  
<http://omim.org/entry/613287>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2O  
<http://omim.org/entry/614228>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2P  
<http://omim.org/entry/614436>
- CHARCOT-MARIE-TOOTH DISEASE, AXONAL, WITH VOCAL CORD PARESIS,  
AUTOSOMAL RECESSIVE  
<http://omim.org/entry/607706>
- CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A  
<http://omim.org/entry/118220>
- CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B  
<http://omim.org/entry/118200>
- CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1C  
<http://omim.org/entry/601098>

- CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1D  
<http://omim.org/entry/607678>
- CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1F  
<http://omim.org/entry/607734>
- CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE B  
<http://omim.org/entry/606482>
- CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE C  
<http://omim.org/entry/608323>
- CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE D  
<http://omim.org/entry/607791>
- CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE E  
<http://omim.org/entry/614455>
- CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE A  
<http://omim.org/entry/608340>
- CHARCOT-MARIE-TOOTH DISEASE, TYPE 4A  
<http://omim.org/entry/214400>
- CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B1  
<http://omim.org/entry/601382>
- CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2  
<http://omim.org/entry/604563>
- CHARCOT-MARIE-TOOTH DISEASE, TYPE 4C  
<http://omim.org/entry/601596>
- CHARCOT-MARIE-TOOTH DISEASE, TYPE 4D  
<http://omim.org/entry/601455>
- CHARCOT-MARIE-TOOTH DISEASE, TYPE 4H  
<http://omim.org/entry/609311>
- CHARCOT-MARIE-TOOTH DISEASE, TYPE 4J  
<http://omim.org/entry/611228>
- CHARCOT-MARIE-TOOTH DISEASE, X-LINKED DOMINANT, 1  
<http://omim.org/entry/302800>
- CHARCOT-MARIE-TOOTH DISEASE, X-LINKED RECESSIVE, 5  
<http://omim.org/entry/311070>
- HEREDITARY MOTOR AND SENSORY NEUROPATHY, TYPE IIC  
<http://omim.org/entry/606071>
- HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS  
<http://omim.org/entry/145900>

- NEUROPATHY, CONGENITAL HYPOMYELINATING OR AMYELINATING, AUTOSOMAL RECESSIVE  
<http://omim.org/entry/605253>
- ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA  
<http://omim.org/entry/180800>

## Sources for This Summary

- Baets J, De Jonghe P, Timmerman V. Recent advances in Charcot-Marie-Tooth disease. *Curr Opin Neurol.* 2014 Oct;27(5):532-40. doi: 10.1097/WCO.0000000000000131. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25110935>
- Bassam BA. Charcot-Marie-Tooth disease variants-classification, clinical, and genetic features and rational diagnostic evaluation. *J Clin Neuromuscul Dis.* 2014 Mar;15(3):117-28. doi: 10.1097/CND.0000000000000020. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24534835>
- OMIM: CHARCOT-MARIE-TOOTH DISEASE, TYPE 4H  
<http://omim.org/entry/609311>
- OMIM: CHARCOT-MARIE-TOOTH DISEASE, TYPE 4J  
<http://omim.org/entry/611228>
- Ekins S, Litterman NK, Arnold RJ, Burgess RW, Freundlich JS, Gray SJ, Higgins JJ, Langley B, Willis DE, Notterpek L, Pleasure D, Sereda MW, Moore A. A brief review of recent Charcot-Marie-Tooth research and priorities. *F1000Res.* 2015 Feb 26;4:53. doi: 10.12688/f1000research.6160.1. eCollection 2015. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25901280>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4392824/>
- El-Abassi R, England JD, Carter GT. Charcot-Marie-Tooth disease: an overview of genotypes, phenotypes, and clinical management strategies. *PM R.* 2014 Apr;6(4):342-55. doi: 10.1016/j.pmrj.2013.08.611. Epub 2014 Jan 13. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24434692>
- GeneReview: Charcot-Marie-Tooth Hereditary Neuropathy Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1358>
- GeneReview: Charcot-Marie-Tooth Neuropathy X Type 5  
<https://www.ncbi.nlm.nih.gov/books/NBK1876>
- Harel T, Lupski JR. Charcot-Marie-Tooth disease and pathways to molecular based therapies. *Clin Genet.* 2014 Nov;86(5):422-31. doi: 10.1111/cge.12393. Epub 2014 May 9. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24697164>
- Hoyle JC, Isfort MC, Roggenbuck J, Arnold WD. The genetics of Charcot-Marie-Tooth disease: current trends and future implications for diagnosis and management. *Appl Clin Genet.* 2015 Oct 19;8:235-43. doi: 10.2147/TACG.S69969. eCollection 2015. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26527893>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4621202/>
- Patzko A, Shy ME. Charcot-Marie-Tooth disease and related genetic neuropathies. *Continuum (Minneapolis Minn).* 2012 Feb;18(1):39-59. doi: 10.1212/01.CON.0000411567.34085.da. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22810069>

- Rossor AM, Polke JM, Houlden H, Reilly MM. Clinical implications of genetic advances in Charcot-Marie-Tooth disease. *Nat Rev Neurol*. 2013 Oct;9(10):562-71. doi: 10.1038/nrneurol.2013.179. Epub 2013 Sep 10. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24018473>
- Saporta MA. Charcot-Marie-Tooth disease and other inherited neuropathies. *Continuum (Minneapolis Minn)*. 2014 Oct;20(5 Peripheral Nervous System Disorders):1208-25. doi: 10.1212/01.CON.0000455885.37169.4c. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25299278>
- Tazir M, Hamadouche T, Nouioua S, Mathis S, Vallat JM. Hereditary motor and sensory neuropathies or Charcot-Marie-Tooth diseases: an update. *J Neurol Sci*. 2014 Dec 15;347(1-2):14-22. doi: 10.1016/j.jns.2014.10.013. Epub 2014 Oct 16. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25454638>
- Vallat JM, Mathis S, Funalot B. The various Charcot-Marie-Tooth diseases. *Curr Opin Neurol*. 2013 Oct;26(5):473-80. doi: 10.1097/WCO.0b013e328364c04b. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23945280>

---

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/charcot-marie-tooth-disease>

Reviewed: December 2015

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services